Health status of children with Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency in the United Kingdom: results of a multi-centre cohort study

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Introduction and Objectives

Congenital adrenal hyperplasia (CAH) is associated with long-term health problems. However, little is known about co-morbidities and their onset in children and young persons (CYP).

Our objective was to establish the health status of CYP with CAH across the United Kingdom.

Conclusion

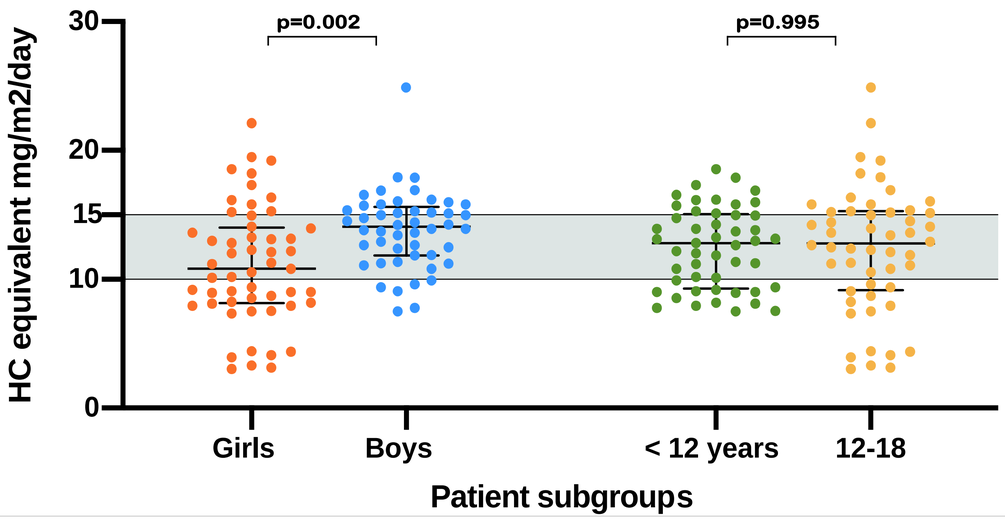
Children with CAH have increased prevalence of growth problems, excessive weight, and metabolic co-morbidities.

The development of improved standardised treatment and personalised strategies for the management and monitoring of CAH in childhood is required in order to improve long-term patient outcomes.

Results

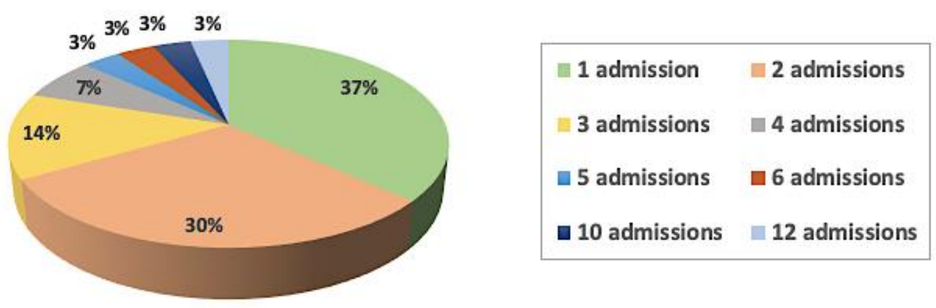
Glucocorticoid treatment (hydrocortisone 94.1%, prednisolone 5.9%) exceeded 15 mg/m^2/day in 27% of patients. 84.3% of patients received 3-4 doses/day with a higher a.m. dose (50-70% of total daily dose in 32% patients; 30-50% of total daily dose in 35%). 75% of patients received fludrocortisone with a median dose 90(64-133) μg/m^2/day.

Figure 1. Glucocorticoid doses in age and gender subgroups
(The shaded area corresponds to the recommended dose range)



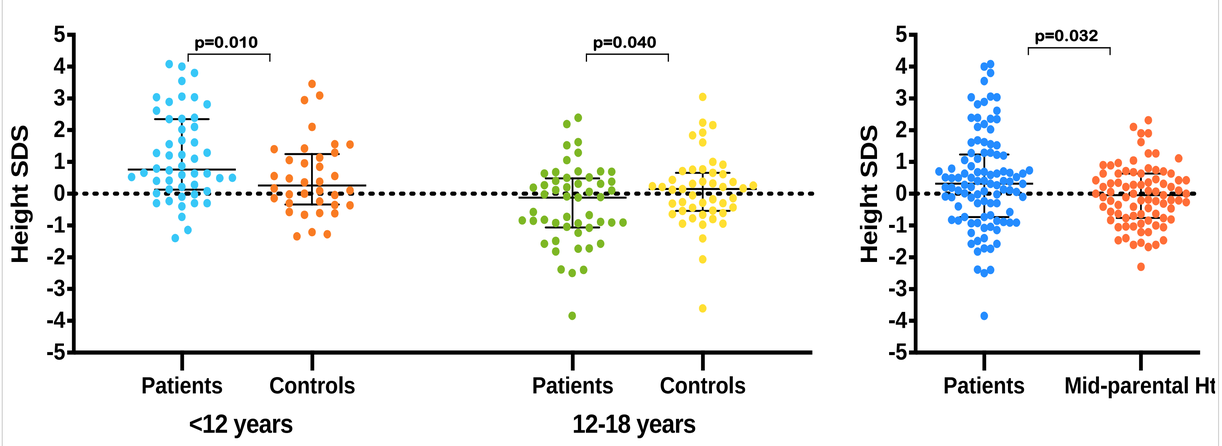
After diagnosis 35 patients (34.3%) required admission for adrenal crisis.

Figure 2. Frequency of admissions for adrenal crisis



Delta-SDS for target height was 1.2±1.4 for children younger than 12 years and 0.3±1.6 for 12-18 year-olds. Comparing height-SDS, patients younger than 12 years were taller (p=0.02) and patients aged 12-18 years shorter (p=0.03) than controls.

Figure 3. Height SDS in patients with CAH compared to healthy controls and parental height



Advanced bone age (>1.5 years) was not different between girls (23%) and boys (20%).

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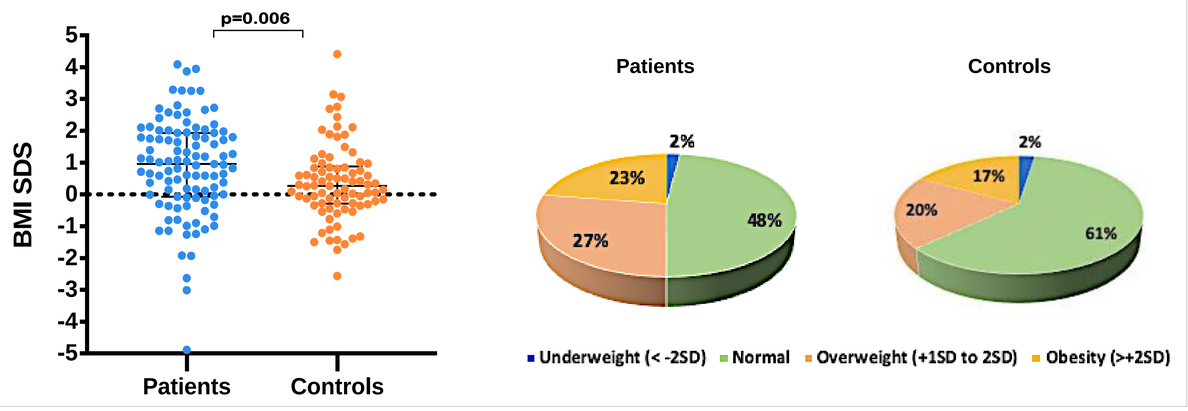
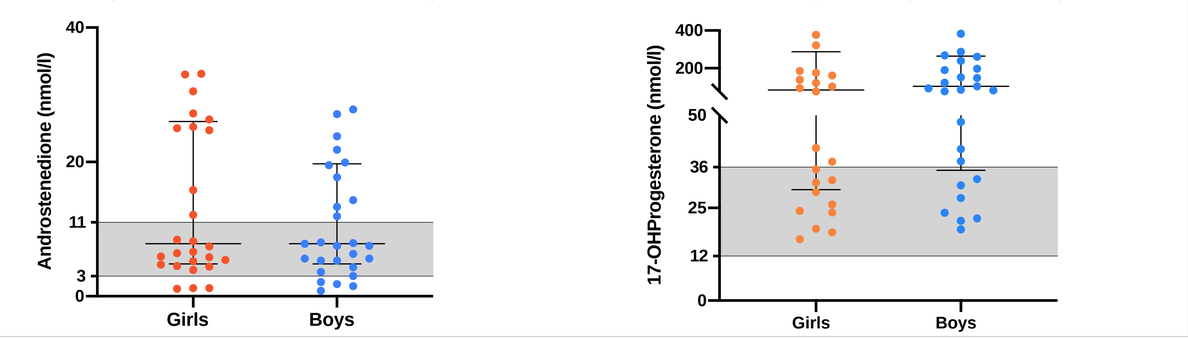
Patient weight-SDS (0.87; 0.03-1.35) and body-mass-index-SDS (0.98; -0.04-1.94) were significantly higher than in controls.

Figure 4. BMI in CAH patients compared to controls



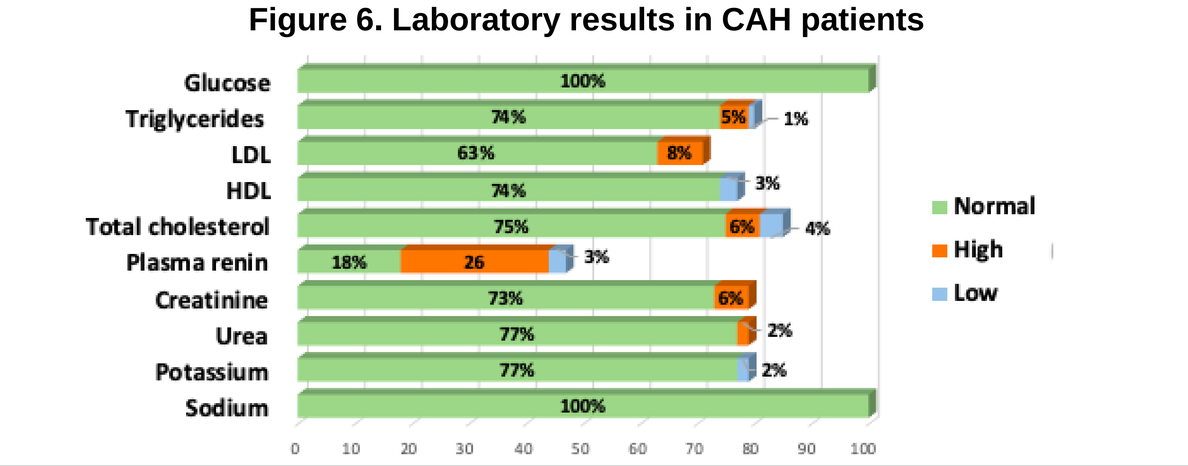
Post-glucocorticoid dose androstenedione was normal in 32%, supressed in 7%, and elevated in 50% cases; 17-hydroxyprogesterone was within target range in 20%, supressed in 19%, and increased in 43% of patients.

Figure 5. Plasma hormones concentrations in patients with CAH
(The shaded areas correspond to the recommended ranges for good control)



One patient presented cushingoid features. Virilisation was reported in seven females. High blood pressure was found in five patients and none of the controls. Plasma biochemistry indicated mildly raised creatinine in 9.8% and abnormal high lipids in 9.8% of CAH patients.

Figure 6. Laboratory results in CAH patients



Associated behavioural and mental health problems were reported for 11.3% patients aged 12-18 years, similar to the general population.

Methods

A multi-centre prospective study recruited 102 patients (54 females) with 21-hydroxylase deficiency targeting CYP aged 8-18 years (13.0±2.92 years) from 13 centres across the United Kingdom and 83 matched controls. Demographic, clinical, and metabolic data were explored by descriptive statistics and analysis of variance.